



succinyl-CoA:3-ketoacid CoA transferase deficiency

Succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency is an inherited disorder that impairs the body's ability to break down ketones, which are molecules produced in the liver during the breakdown of fats.

The signs and symptoms of SCOT deficiency typically appear within the first few years of life. Affected individuals experience episodes of extreme tiredness (lethargy), appetite loss, vomiting, rapid breathing, and, occasionally, seizures. These episodes, which are called ketoacidotic attacks, sometimes lead to coma. About half of affected individuals have a ketoacidotic attack within the first 4 days of life. Affected individuals have no symptoms of the disorder between ketoacidotic attacks.

People with SCOT deficiency usually have a permanently elevated level of ketones in their blood (persistent ketosis). If the level of ketones gets too high, which can be brought on by infections, fevers, or periods without food (fasting), a ketoacidotic attack can occur. The frequency of ketoacidotic attacks varies among affected individuals.

Frequency

The prevalence of SCOT deficiency is unknown. More than 20 cases of this condition have been reported in the scientific literature.

Genetic Changes

Mutations in the *OXCT1* gene cause SCOT deficiency. The *OXCT1* gene provides instructions for making an enzyme called succinyl-CoA:3-ketoacid CoA transferase (SCOT). The SCOT enzyme is made in the energy-producing centers of cells (mitochondria). The enzyme plays a role in the breakdown of ketones, which are an important source of energy during fasting or when energy demands are increased, such as during illness or when exercising.

OXCT1 gene mutations result in the production of a SCOT enzyme with little or no function. A reduction in the amount of functional enzyme leads to an inability to break down ketones, resulting in decreased energy production and an elevated level of ketones in the blood. If these signs become severe, a ketoacidotic attack can occur. Individuals with mutations that create an enzyme with partial function are still prone to ketoacidotic attacks, but are less likely to have persistent ketosis.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal

recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- 3-oxoacid CoA transferase deficiency
- ketoacidosis due to SCOT deficiency
- SCOT deficiency
- succinyl-CoA 3-oxoacid transferase deficiency
- succinyl-CoA:3-oxoacid CoA transferase deficiency
- succinyl-CoA:acetoacetate transferase deficiency

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Succinyl-CoA acetoacetate transferase deficiency
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0342792/>

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Ketones--Urine
<https://medlineplus.gov/ency/article/003585.htm>
- MedlinePlus Encyclopedia: Serum Ketones Test
<https://medlineplus.gov/ency/article/003498.htm>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Ketones--Urine
<https://medlineplus.gov/ency/article/003585.htm>
- Encyclopedia: Serum Ketones Test
<https://medlineplus.gov/ency/article/003498.htm>
- Health Topic: Mitochondrial Diseases
<https://medlineplus.gov/mitochondrialdiseases.html>

Genetic and Rare Diseases Information Center

- SCOT deficiency
<https://rarediseases.info.nih.gov/diseases/4774/scot-deficiency>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Seizures and Epilepsy
<https://www.ninds.nih.gov/Disorders/All-Disorders/Epilepsy-Information-Page>

Educational Resources

- CLIMB: Succinyl-CoA 3-Oxoacid Transferase Deficiency Info Sheet
<http://www.climb.org.uk/IMD/Sierra/Succinyl-CoA3-OxoacidTransferaseDeficiency.pdf>
- Disease InfoSearch: SCOT deficiency
<http://www.diseaseinfosearch.org/SCOT+deficiency/6468>
- Disease InfoSearch: Succinyl-CoA acetoacetate transferase deficiency
<http://www.diseaseinfosearch.org/Succinyl-CoA+acetoacetate+transferase+deficiency/6918>
- MalaCards: succinyl coa:3-oxoacid coa transferase deficiency
http://www.malacards.org/card/succinyl_coa3_oxoacid_coa_transferase_deficiency_2
- Orphanet: Succinyl-CoA:3-ketoacid CoA transferase deficiency
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=832

Patient Support and Advocacy Resources

- CLIMB: Children Living with Inherited Metabolic Diseases
<http://www.climb.org.uk/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28scot+deficiency%5BTIAB%5D%29+OR+%28succinyl-coa:3-ketoacid+coa+transferase%5BTIAB%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- SUCCINYL-CoA:3-OXOACID-CoA TRANSFERASE DEFICIENCY
<http://omim.org/entry/245050>

Sources for This Summary

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Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/succinyl-coa3-ketoacid-coa-transferase-deficiency>

Reviewed: December 2011

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services